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Letter to the Editor

Inheritance of Niikawa-Kuroki (Kabuki Makeup) Syndrome

To the Editor:

Kobayashi and Sakuragawa [1996] recently reported on a familial case of Niikawa-Kuroki (Kabuki make-up) syndrome, suggesting autosomal-dominant inheritance. Another family demonstrating autosomal-dominant inheritance was described by Halal et al. [1980]. In both reports, significant intrafamilial variability in the clinical expression of the syndrome was evident. In both pedigrees, the fathers were the mildly affected, transmitting carriers. Neither had short stature or mental retardation.

Kobayashi and Sakuragawa [1996] suggested that only individuals with milder symptoms are likely to reproduce, so that the disease may be transmitted to a second generation where the more severe manifestations occur.

An ascertainment bias due to underdiagnosis of milder cases might account for the apparently sporadic occurrence of most of the over 100 reported cases.

We would like to add a personal contribution to support autosomal-dominant inheritance and variable clinical expressivity of Niikawa-Kuroki syndrome. A previously reported Italian girl with the typical findings of Niikawa-Kuroki syndrome had a mother with minor facial anomalies and hypoplastic root of a premolar on Panorex [Silengo et al., 1991]. Interestingly, the mildly affected father in the Halal family did have hypodontia of the premolars [Halal et al., 1980]; tooth anomalies are a common finding in Niikawa-Kuroki syndrome, being observed in over 70% of patients.

The affected mother in our family had a termination of pregnancy because of sonographic evidence of diaphragmatic hernia in the fetus.

A review of non-Japanese sporadic cases of Niikawa-Kuroki syndrome by Philip et al. [1992] showed the presence of a diaphragmatic defect in 2 of 17 patients.

We suggest that the diaphragmatic hernia in the aborted fetus represents an uncommon manifestation of Niikawa-Kuroki syndrome, further supporting autosomal-dominant inheritance in our family.

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